Medical and Pediatric Oncology 30:67–68 (1998)

Poland's Syndrome and Wilms Tumor: An Unusual Association

Uma H. Athale, MD,* and Rajasekharan Warrier, MD

Poland's syndrome, a rare congenital disorder with pectoralis muscular girdle defect, have been reported in association with lymphoreticular malignancies in the past. Childhood solid tumors in association with this congenital anomaly have not been reported so far. We describe this rare association of Poland's syndrome and Wilms tumor. Due to the possibility of increased risk of leukemogenesis in patients with Poland's syndrome, chemo-radiation therapy of Wilms tumor in our patient may increase the risk of secondary leukemia. Therapeutic modification of primary cancer in these patients may be necessary with careful long-term follow-up for early detection and treatment of secondary cancer. Med. Pediatr. Oncol. 30:67–68, 1998. © 1998 Wiley-Liss, Inc.

Key words: Wilms tumor; Poland's syndrome; leukemia; solid tumor; secondary cancer

INTRODUCTION

Poland's Syndrome is a rare congenital disorder characterized by aplasia of pectoralis major muscle and varying degrees of ipsilateral upper limb deficiency [1]. Wilms tumor has been associated with several congenital anomalies, noteworthy of them being Beckwith-Weidmann Syndrome, hemihypertrophy, aniridia, and genitourinary anomalies [2]. Poland's Syndrome, however, has not so far been reported in patients with Wilms tumor.

We report an unusual association of Poland's Syndrome and Wilms tumor.

CASE REPORT

J.H., a 17-month-old white boy, diagnosed to have Poland's syndrome was referred to us for a renal mass which was detected on abdominal ultrasound examination done by the referring pediatrician to rule out associated renal anomalies. He was born of a nonconsanguineous marriage of normal pregnancy and delivery and the family did not report any congenital anomalies and/or cancer in the immediate family members.

Physical examination revealed a comfortable boy with weight of 11.7 kg (25th percentile for age) and height of 80 cm (50th percentile for age). The congenital skeletal anomalies included absence of sternal head of right pectoralis major muscle with small ipsilateral pectoralis minor; breast hypoplasia with superior placement of right nipple and shortening of right upper limb with syndactyly of right second and third digit. In addition, he also had shortening of right lower limb. He had a palpable nontender left renal mass. He had normal male genitalia with bilateral descended testes. He was hypertensive with blood pressure of 157/86 mm of mercury. The rest of his examination was normal.

The urinalysis, hemogram, blood chemistry, and coagulation profile were normal for age. He had a normal chest x-ray and normal skeletal survey. CT scan of the chest showed normal lung parenchyma free from any metastatic disease. The right chest wall was thinner than the left side which was consistent with the diagnosis of Poland's Syndrome. Abdominal ultrasound showed a large solid mass of 5 cm diameter arising from the upper pole of the left kidney which was consistent with Wilms tumor. Right kidney was unremarkable and measured 6.2 cm. There was no evidence of genitourinary anomaly, venous extension or intra-abdominal spillage of the tumor. EKG and ECHO/Doppler studies were within normal limits.

With the clinical diagnosis of variant of Poland's anomaly and stage I Wilms tumor the patient underwent left radical nephroureterectomy and simultaneous central line placement. He tolerated the surgery well.

Cytogenetic analysis of both peripheral blood and tumor tissue revealed a normal male karyotype (46XY) with normal 11p. DNA analysis showed a DNA index of 1.0; and diploid cell population. No aneuploidy was detected.

Microscopic examination confirmed diagnosis of favorable histology Wilms tumor confined to the left kidney with free surgical margins. Tumor cells were noted in the peritumoral vessels, vessels in the renal mass and

Department of Pediatric Hematology/Oncology, Louisiana State University School of Medicine and Children's Hospital, New Orleans, LA.

*Correspondence to: Dr. Uma H. Athale, Department of Pediatric Hematology/Oncology, Louisiana State University School of Medicine, 1542 Tulane Avenue, New Orleans, LA 70118.

Received 17 April 1996; Accepted 28 June 1996

also in the subcapsular lymphatic space of one of the right paraparetic lymph nodes.

On the basis of clinical and pathological features he was staged as Wilms tumor, favorable histology Stage III.

He was started on National Wilms tumor study 4 protocol with chemotherapy (consisting of Vincristine, Adriamycin, and Actinomycin D for 65 weeks) and postoperative radiation therapy with 1080 cGy delivered in six fractions to the left flank consisting of the renal bed and tumor volume with 1–2 cms margin extending across the midline, including the para-aortic lymph nodes.

DISCUSSION

Alfred Poland first described the association of ipsilateral pectoralis girdle muscular deficiency and syndactyly in a 27-year-old convict [1]. Others have further described the several variants of Poland's syndrome and other associated congenital anomalies including primary microcephaly, peroneal muscular atrophy, spinal and genitourinary defects [3–6]. In addition to the classical constellation of absent pectoralis major, and ipsilateral limb defects and hypoplasia of breast; our patient had shortening of ipsilateral lower limb which makes it a variant of Poland's syndrome. His contralateral anthropometric measurements were at the 50th percentile for his age ruling out hemihypertrophy of the opposite side.

The exact etiology of Poland's anomaly is unknown. But, interference in subclavian arterial blood supply during the early embryonic period is postulated [3,4]. An alternative hypothesis presumes it to be secondary to an early local defect in the mesoderm leading to incomplete development of the structures involved [3]. Although no cytogenetic abnormality have been so far reported with Poland's syndrome, an as yet unidentified genetic or chromosomal defect cannot be ruled out especially on the background of several reported familial occurrences of this defect [7].

Poland's syndrome has been reported in association with hematological malignancies like acute leukemias (both acute lymphoblastic and acute nonlymphoblastic) and chronic granulocytic leukemia; non-Hodgkin's lymphoma [8–12]. These reports suggest that patients with Poland's syndrome may have an increased predisposition to leukemia. The biological basis for the lymphoreticular malignancies in association with Poland's syndrome is unclear [12]. An unidentified genetic or chromosomal defect, as suggested earlier, may predispose these patients to cancers. A further in depth study is needed.

Leiomyosarcoma in a 56-year-old woman with Poland's syndrome is the only solid malignant tumor that has been reported so far in association with this congenital anomaly [13]. Our case is the first reported occurrence of Wilms tumor with Poland's anomaly. The occurrence of solid tumor in our patient with Poland's syndrome may be a reflection of the possible predisposition to cancer or a mere chance occurrence.

Most of the reported cases with Poland's syndrome and leukemia were children below 14 years. The available data is inadequate to evaluate the prognosis and outcome of leukemia in Poland's syndrome. The two-year disease-free survival of isolated Stage III Wilms tumor with favorable histology is in the range of 90 percent [14]. Any effect on prognosis due to the interaction of Poland's syndrome (and underlying unidentified biological aberration) with Wilms tumor is unknown.

Due to the possible predisposition to malignancy, persons with Poland's syndrome need to be carefully monitored for early detection and therapy of cancer. Therapeutic modification in treatment of primary cancer may be necessary to avoid development of secondary cancer.

REFERENCES

1. Poland A. Deficiency of the pectoralis muscle. Guy's Hospital Reports 6:191–193, 1841.
2. Mesrobian HGJ. Wilms' tumor: past, present, and future. J Urol 140:231–238, 1988.
3. Wilson MR, Louis DS, Stevenson TR. Poland's syndrome: variable expression and associated anomalies. J Hand Surg 6:880–2, 1988.
4. Fryns JP, de Smet L. On the association of Poland anomaly and primary microcephaly. Clin Dysmorphol 3:347–50, 1994.
5. Asaka T. Peroneal muscular atrophy with Poland's syndrome. Acta Neurol Scand 89:307–8, 1995.
6. Fabian MC, Fischer JD. A variant of Poland's syndrome. Can J Surg 37:67–9, 1994.
7. Soltan HC, Holmes LB. Familial occurrence of malformations possibly attributable to vascular abnormalities. J Pediatr 108:112–4, 1986.
8. Boaz D, Mace JW, Gotlin RW. Poland's Syndrome and Leukemia. Lancet 1:1349–50, 1971.
9. Sackey K, Odone V, George S, Murphy S. Poland's syndrome associated with childhood non-Hodgkin's lymphoma. American Journal of Diseases of Childhood 138:600–1, 1984.
10. Parikh PM, Karandikar SM, Koppikar S, Pahuja R, Charak BS, Saikia T, Gopal R, Advani SH. Poland's syndrome with acute lymphoblastic leukemia in an adult. Med Pediatr Oncol 16:290–2, 1988.
11. Costa R, Afonso E, Benedito M, Maricato L. Poland's syndrome associated with chronic granulocytic leukemia. Sangre 36:417–8, 1991.
12. Gilman PA, Miller RW. No link between Poland's syndrome and leukemia? Letter. The American Journal of Diseases of Childhood 176, 1982.
13. Shaham D, Ramu H, Bar-Ziv J. Leiomyosarcoma in Poland's syndrome. A case report. Acta Radiol 33:444–6, 1992.
14. D'Angio GJ, Breslow N, Beckwith JB, et al. The treatment of Wilms' tumor: Results of the Third National Wilms' Tumor Study. Cancer 64:466–479, 1989.